

The Human Genome Project is an ongoing collaborative effort to unravel human DNA and understand the genetic code of life. Already more than 10,000 genes have been discovered and hundreds of genetic tests developed for commercial use. Now a major milestone has been reached—the first draft of the human genome sequence.

A Public Health Perspective

The Human Genome Project: “Gene Sequencing and Discovery are only the Beginning”

*Muin J. Khoury MD, PhD
Director, Office of Genetics and Disease Prevention
Centers for Disease Control and Prevention*

Excitement and Confusion



Excitement about the progress of the Human Genome Project abounds in both research and medical communities. However, as excitement abounds, so does confusion. While many people worry about social and ethical issues surrounding genetics, others say that a revolution in medicine is occurring—a revolution that will usher in a new era of disease prevention and treatment.

Although there appears to be agreement that the impact of the Human Genome Project will probably be felt across all disease areas, a basic question remains. What do the genetic discoveries of the Human Genome Project mean to disease prevention and good health?

Only the Beginning

It could mean plenty, according to the Office of Genetics and Disease Prevention at the Centers for Disease Control and Prevention (OGDP/CDC). However, gene discovery is only the beginning. For most diseases, a wide gap exists between sequencing and discovering genes and the safe and effective use of genetic information to prevent disease and improve health. OGDG/CDC is working with other members of the public health community to narrow this gap, but the task is enormous and is just beginning.

Beyond Gene Discovery

Moving beyond gene discovery to relevant action in health care will require additional research and careful planning by public health officials. Medical and public health researchers must evaluate in

The Human Genome Project: "Gene Sequencing and Discovery are only the Beginning" Continued

different populations each of the thousands of genes and gene variations to

1. Determine the relationship between genetic variation and risks for various diseases
2. Identify gene variations and other risk factors, such as behavior and environment, that may interact with genes
3. Evaluate the usefulness and accuracy of genetic tests and the utility of interventions in preventing disease and improving health
4. Assess the impact of genetic tests and other services on individuals, families, and society
5. Address associated social, legal, and ethical issues

A Solid Foundation

As these evaluation steps are completed, our knowledge of disease processes, the complex interplay of genes and environment, and the risk for disease is also increasing. Future intervention strategies—medical, behavioral, or otherwise—will be based on Human Genome Project data that have been validated with public health research. Accurate and meaningful genetic information will be available to create a solid foundation for public health policy and programs.

A New Era in Medicine

The Human Genome Project will be heralded as one of the most astounding medical projects of all time and will provide the foundation for a new era in medicine. The next step will be to integrate genetics into all aspects of health policy through applied medical and public health research, educating and training the workforce, and assuring access to quality genetic information. Our challenge is to use genetic information safely and effectively to prevent disease and improve health for the citizens of the 21st century.